

EFFECTIVE
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NEBRASKA HEALTH AND HUMAN SERVICES
REGULATION AND LICENSURE

181 NAC 5

TITLE 181 SPECIAL HEALTH PROGRAMS

CHAPTER 5 CONSENT FOR PREDICTIVE GENETIC TESTING

5-001 SCOPE AND AUTHORITY: These regulations implement Neb. Rev. Stat. § 71-1,104.01 governing informed consent for predictive genetic testing. These regulations define terms, state the requirements for use of the model informed consent form, provide the model informed consent form, and specify the conditions under which use of the model form confers immunity from civil liability.

5-002 DEFINITIONS

Department means the Department of Health and Human Services Regulation and Licensure.

Genetic information means information about a gene, gene product, or inherited characteristic derived from a genetic test.

Genetic test means analysis of human DNA, RNA, chromosomes, epigenetic status, and those tissues, proteins, and metabolites used to detect heritable or somatic disease-related genotypes or karyotypes for clinical purposes. Tests of tissues, proteins, and metabolites are included only when generally accepted in the scientific and medical communities as being specifically determinative of a heritable or somatic disease-related genetic condition. *Genetic test does not include a routine analysis, including a chemical analysis, of body fluids or tissues unless conducted specifically to determine a heritable or somatic disease-related genetic condition. Genetic test does not include a physical examination or imaging study.*

Model informed consent form means the portion of the form attached to and incorporated in these regulations which pertains to the type of predictive genetic testing being offered. Attachment A is the supplemental newborn screening consent form, Attachment B is the predictive prenatal genetic testing consent form, and Attachment C is the predictive genetic testing consent form.

Patient representative includes –

1. The custodial parent or guardian of an unemancipated minor, except that –
 - a. A minor who is pregnant has authority to give informed consent or refusal for pregnancy-related medical procedures unless, due to physical or intellectual limitations, she is unable to make and communicate a considered judgment about her medical care; and
 - b. The minor parent of a child has authority to give informed consent or refusal for the medical care of his or her child unless the parent, due to physical or intellectual limitations, is unable to make and communicate a considered judgment about the child's medical care.
2. The legal guardian, attorney in fact appointed by a Durable Power of Attorney for Health Care, or other adult who has a history of assuming decisional authority for a patient who, due to physical or intellectual limitations, is unable to make and communicate a considered judgment about his or her medical care.

Physician means a person licensed to practice medicine and surgery or osteopathic medicine and surgery pursuant to Neb. Rev. Stat. §§ 71-1,102 to 71-1,107.30, 71-1,137 to 71-1,141, and the Uniform Licensing Law.

Physician delegate means an individual acting under the delegated authority of a physician to perform a selected act, task, or function, and who understands and is qualified to provide the information required in the written informed consent.

Predictive genetic test means a genetic test for an otherwise undetectable genotype or karyotype relating to the risk for developing a genetically related disease or disability, the results of which can be used to substitute a patient's prior risk based on population data or family history with a risk based on genotype or karyotype. *Predictive genetic test does not include diagnostic testing conducted on a person exhibiting clinical signs or symptoms of a possible genetic condition. Predictive genetic testing does not include gamete testing, preimplantation diagnosis, or prenatal genetic diagnosis, unless the prenatal testing is conducted for an adult-onset condition not expected to cause clinical signs or symptoms before the age of majority.*

Predictive prenatal genetic test means a test of fetal tissue, chorionic villi, or amniotic fluid for an adult-onset condition not expected to cause clinical signs or symptoms before the age of majority. Predictive prenatal genetic testing does not include maternal or fetal screening or testing for conditions expected to cause clinical signs or symptoms before the age of majority.

Required newborn screening means those newborn screening tests required by Neb. Rev. Stat. § 71-519. This also includes the required newborn screening panel from the birth state for newborns born in other states but screened in Nebraska.

Supplemental newborn screening means those tests which may be ordered for newborns which are not required by law but may be made available to the newborn and which meet the definition of predictive genetic test.

Written informed consent means a signed writing executed by the patient or patient representative which confirms that the physician or physician delegate has explained, and the patient or patient representative understands –

1. The nature and purpose of the predictive genetic test;
2. The effectiveness and limitations of the predictive genetic test;
3. The implications of taking the predictive genetic test, including the medical risks and benefits;
4. The future uses of the sample taken to conduct predictive genetic test and the genetic information obtained from the predictive genetic test;
5. The meaning of the predictive genetic test results and the procedure for providing notice of the results to the patient; and
6. Who will have access to the sample taken to conduct the predictive genetic test and the genetic information obtained from the predictive genetic test, and the patient's right to confidential treatment of the sample and the genetic information.

5-003 PHYSICIAN DUTIES

5-003.01 With the exception of required newborn screening, a physician or the physician's delegate must obtain written informed consent from the patient or the patient's representative before ordering a predictive genetic test. The ordering physician or physician delegate must be reasonably well qualified to provide the information required.

5-003.02 The discussion process leading to informed consent must include the elements listed under the definition of "written informed consent" in 181 NAC 5-002. The written informed consent document must indicate that these elements have been explained by the physician or physician delegate and understood by the patient or patient representative. Use of the model informed consent form is not required, but if the model form is properly completed and is signed by the patient or patient representative, the patient is barred from bringing a civil action for damages against the physician or physician delegate based on failure to obtain informed consent for the predictive genetic test. Use of the model informed consent form will not protect against possible liability for the improper testing of a minor.

5-003.03 Required newborn screening tests are exempted from the requirement for written informed consent, but the attending physician must inform the parent about the required tests. Supplemental newborn screening is not exempted and requires written informed consent. Providing the parent with the written materials on required newborn screening prepared by the Department and proper completion of Attachment A will satisfy both requirements.

5-003.04 The requirement for written informed consent prescribed by 181 NAC 5 applies only to predictive genetic testing relating to an asymptomatic individual's risk of developing a genetically related disease or disability. Proper completion of Attachment B will satisfy this obligation when ordering predictive prenatal genetic testing for an adult-onset disorder. Proper completion of Attachment C will satisfy this obligation for all predictive genetic tests other than predictive prenatal genetic testing and supplemental newborn screening.

5-003.04A Written informed consent is required before ordering any of the following tests on a patient without clinical signs or symptoms –

1. Human DNA, RNA, epigenetic, and chromosomal tests used to detect heritable or somatic disease-related genotypes or karyotypes for clinical purposes;
2. Tissue, protein and metabolic tests when generally accepted in the scientific and medical communities as being specifically determinative of a heritable or somatic disease-related genetic condition;
3. Prenatal genetic tests or predictive genetic tests of a minor for an adult-onset genetic condition(s) not expected to cause clinical signs or symptoms before the age of majority; and
4. Tests which can be used to alter risk assessment by substituting a genotype or karyotype-based risk for a risk based on population data or family history.

5-003.04B Because the following acts and procedures do not fall under the definition of predictive genetic testing for purposes of these regulations (see 5-002), written informed consent is not required pursuant to these regulations for –

1. Diagnostic testing in a patient showing clinical signs or symptoms of a possible genetic condition;
2. Testing for carrier status of a recessive gene, balanced translocation, or other genetic marker in a person not expected to develop clinical signs or symptoms due to carrier status;
3. Gamete testing or preimplantation genetic diagnosis;
4. Prenatal genetic testing or screening, unless the test is for an adult-onset genetic condition not expected to cause clinical signs or symptoms before the age of majority;
5. Routine analysis (such as blood typing) not typically associated with genetic disease or disability and not performed with the intention of assessing the patient's risk for future genetic disease or disability;
6. Research protocols for which Institutional Review Board-approved consent forms must be used;
7. Physical examinations; and
8. Imaging studies.

5-003.05 Copy of Form: A copy of the completed written informed consent must be provided to the patient or patient's representative and the original must be filed in the patient's medical record.

5-003.06 Use of Model Informed Consent Forms

5-003.06A Attachment A, consent for supplemental newborn screening, is to be used only for supplemental newborn screening. Written educational materials on newborn screening provided by the Department must accompany it. To be considered properly completed, the following information must appear on the form –

1. The baby's name, date of birth, and ID number;
2. The name and phone number of the person to be called in the event of questions;
3. The name of the reference laboratory where the specimen will be sent;
4. The physician or physician's delegate's signature and date; and
5. The patient or patient's representative's signature and date.

If the supplemental screening is declined, the patient or patient representative may do so in writing, as provided on Attachment A.

5-003.06B Attachment B, consent for predictive prenatal genetic testing, is to be used only when ordering a predictive prenatal genetic test for an adult-onset genetic condition not expected to cause clinical signs or symptoms before the age of majority. To be considered properly completed, the following information must appear on the form –

1. Checkmarks in all boxes applicable to the proposed test;
2. Completion of all blanks applicable to the proposed test;
3. The name(s) of the reference laboratory or laboratories to which the sample will be sent;
4. The manner in which results are to be received if testing is accepted;
5. The signature and date of signature of the patient or patient representative if testing is accepted;
6. The signature and date of signature of the physician or physician delegate who conducted the discussion process; and
7. The name and phone number of the person to be called in the event of questions.

If testing is declined, the patient or patient representative may do so in writing, as provided on Attachment B.

5-003.06C Attachment C, consent for predictive genetic testing, is to be used for all tests meeting the definition of predictive genetic testing other than required newborn screening, supplemental screening, and predictive prenatal genetic testing. To be considered properly completed, the following information must appear on the form –

1. Checkmarks in all boxes applicable to the proposed test;
2. Completion of all blanks applicable to the proposed test;
3. The name(s) of the reference laboratory or laboratories to which the sample will be sent ;
4. The manner in which results are to be received if testing is accepted;
5. The signature and date of signature of the patient or patient representative if testing is accepted;
6. The signature and date of signature of the physician or physician delegate who conducted the discussion process;
7. The name and phone number of the person to be called in the event of questions.

If testing is declined or deferred to a later time, the patient or patient representative may do so in writing, as provided on Attachment C.

MODEL CONSENT FORM FOR SUPPLEMENTAL NEWBORN SCREENING

General Information about Newborn Screening in Nebraska

Nebraska law says that every newborn **MUST** be tested for certain disorders (described in the state's newborn screening brochure) that could lead to serious problems if they are not found and treated right away. The Nebraska Department of HHS Regulation & Licensure manages this program. We will be testing your baby for these conditions, using blood from your baby's heel. We prick the heel and squeeze a few drops of blood onto a piece of paper. There is a tiny chance of prolonged bleeding if your baby's blood does not clot normally or if your baby is on a blood thinner. Your baby may get a small bruise. There is a small chance of infection.

As with all laboratory tests, there is a slight chance for error. Sometimes samples are damaged in transport. Sometimes samples are labeled wrong, the equipment doesn't run correctly or the results are not interpreted correctly. The chance this will happen, and escape detection, is small because safeguards are built in.

The sample will be sent to a special laboratory. No one else will have access to it. The laboratory will keep the sample for 3 months in case the test needs to be repeated, then it will destroy the sample. Test results will be kept by the laboratory; the hospital or clinic where your baby's blood was drawn; your doctor's office; and the State Department of HHS Regulation & Licensure. Sometimes parents may be asked to donate their baby's sample for research after testing is complete. If that happens, a separate permission form will be used for the donation.

This is a first-step, screening test. Your doctor will call you if more testing is needed to see if your baby really has one of these conditions.

You or your insurance company will be billed for the tests.

Your baby's sample will be sent to the lab contracting with the State Department of HHS Regulation & Licensure to perform the screening: _____

(Name of Laboratory)

NEBRASKA REQUIRED NEWBORN SCREENING

The conditions for which screening is required are:

- *PKU (phenylketonuria) -- requires special diet*
- *galactosemia -- requires special diet*
- *biotinidase deficiency -- requires daily vitamin supplement*
- *congenital primary hypothyroidism -- requires daily medicine*
- *hemoglobin variations -- may require medicine and other care*
- *MCAD -- requires special attention during illness*
- *congenital adrenal hyperplasia -- requires daily medicine* (for specimens received at the newborn screening laboratory on or after January 2, 2006)
- *Cystic fibrosis -- requires daily medicine & special diet* (for specimens received at the newborn screening laboratory on or after January 2, 2006)

SUPPLEMENTAL NEWBORN SCREENING

When we send your baby's blood sample to be tested for MCAD and PKU, the sample goes through a machine that can test many different things in your baby's blood. These results can show other disorders which are NOT on the state's required testing list. The supplemental screening results will only be done if you give permission. If you decide against supplemental screening and later change your mind, a new blood sample will be needed, and new fees will be charged.

The types of disorders found by the supplemental screening are described at the end of this form. These disorders are rare, and they can be hard for your baby's doctor to diagnose. This screening test does not show all possible disorders, but it covers about 30 conditions. Their names are listed in the state newborn screening brochure.

Here are some reasons people might want to know the additional information:

some disorders can cause poor growth
some disorders can cause sudden, very serious illness, permanent disability, or death
without screening, it can be slow and expensive to figure out what's wrong
many disorders can be treated, making the illness less severe or less frequent

the information could be important to other family members

baby's name _____
date of birth _____
ID number _____

Here are some reasons people might not want to know the additional information:

*some treatments are not proven
some disorders may cause death or disability despite medical help
some disorders cause problems for only a few people who have them
sometimes we can't tell whether a pattern is normal or abnormal
final testing may show that the screening result was a false alarm
rarely, the screening test will be normal when the child actually has a disorder
some people worry about possible discrimination in employment or insurance*

This is a first-step, screening test. Your doctor will call you if more testing is needed to see if your baby really has one of these disorders. The screening results should be back in 5-10 days.

You will receive a copy of this form. If you have questions later, call this person:
_____ at this number: _____.

Accept Supplemental Screening Results

After reviewing this information, I have decided to receive the supplemental screening test.

parent, guardian or representative date physician or physician's delegate

Decline Supplemental Screening Results

After reviewing this information, I have decided NOT to receive the supplemental screening test.

parent, guardian or representative date physician or physician's delegate

Description of disorders in the Supplemental Screening Test Panel

The supplemental screen includes the following types of disorders. The names of the conditions are listed in the state newborn screening brochure.

Fatty acid disorders. People with these disorders cannot turn fat into energy in the normal way. They may get low blood sugar, muscle weakness, very low energy, seizures, coma, liver damage, permanent mental or physical disability, or death. Problems may appear in infancy, childhood, adulthood, only during illness, or never.

Amino acid disorders. People with these disorders cannot use certain proteins in the normal way. They may have mental retardation, developmental disabilities, seizures, blindness, or early death.

Organic acid disorders. People with these disorders have trouble using proteins and fats in the normal way. They may get any of the symptoms listed in "fatty acid disorders" and "amino acid disorders."

DEFINITIONS AND USE OF NEBRASKA'S *MODEL* INFORMED CONSENT FORM FOR PREDICTIVE PRENATAL GENETIC TESTING FOR AN ADULT ONSET CONDITION NOT EXPECTED TO CAUSE SIGNS OR SYMPTOMS BEFORE THE AGE OF MAJORITY

Nebraska law requires written, informed consent from the patient or the patient's representative before a predictive genetic test is ordered. Prenatal diagnosis and screening are excluded from the definition of predictive genetic testing unless the prenatal test is for an adult-onset genetic condition. This consent form is intended **ONLY** for patients considering invasive prenatal diagnosis for an adult-onset genetic condition not expected to cause signs or symptoms before the age of majority.

For purposes of this requirement:

Genetic test means analysis of human DNA, RNA, chromosomes, epigenetic status, and those tissues, proteins and metabolites used to detect heritable or somatic disease-related genotypes or karyotypes for clinical purposes. Tests of tissues, proteins and metabolites are included only when generally accepted in the scientific and medical communities as being specifically determinative of a heritable or somatic disease-related genetic condition. *Genetic test does not include a routine analysis, including a chemical analysis, of body fluids or tissues unless conducted specifically to determine a heritable or somatic disease-related genetic condition. Genetic test does not include a physical examination or imaging study.*

Patient representative includes:

- (1) the custodial parent or guardian of an unemancipated minor, except that:
 - (a) a minor who is pregnant has authority to give informed consent or refusal for pregnancy related medical procedures unless, due to physical or intellectual limitations, she is unable to make and communicate a considered judgment about her medical care; and
 - (b) the minor parent of a child has authority to give informed consent or refusal for the medical care of his or her child unless the parent, due to physical or intellectual limitations, is unable to make and communicate a considered judgment about the child's medical care.
- (2) the legal guardian, attorney in fact appointed by a Durable Power of Attorney for Health Care, or other adult who has a history of assuming decisional authority for a patient who, due to physical or intellectual limitations, is unable to make and communicate a considered judgment about his or her medical care.

Physician delegate means an individual acting under the delegated authority of a physician to perform a selected act, task or function, and who understands and is qualified to provide the information required in the written informed consent.

Predictive genetic test means a genetic test for an otherwise undetectable genotype or karyotype relating to the risk for developing a genetically related disease or disability, the results of which can be used to substitute a patient's prior risk based on population data or family history with a risk based on genotype or karyotype. *Predictive genetic test does not include diagnostic testing conducted on a person exhibiting clinical signs or symptoms of a possible genetic condition. Predictive genetic testing does not include gamete testing, pre-implantation diagnosis, or prenatal genetic diagnosis, unless the prenatal genetic diagnosis is conducted for an adult-onset condition not expected to cause clinical signs or symptoms before the age of majority.*

Predictive prenatal genetic test means a test of fetal tissue, chorionic villi or amniotic fluid for an adult-onset condition not expected to cause clinical signs or symptoms before the age of majority. *Predictive prenatal genetic testing does not include gamete testing or pre-implantation diagnosis. Predictive prenatal genetic testing does not include maternal or fetal screening or testing for conditions expected to cause clinical signs or symptoms before the age of majority.*

When properly completed and signed, this form gives evidence that the physician (or physician delegate) and the patient (or patient representative) have thoroughly discussed the risks, benefits and limitations of the proposed test. Patients who have signed this properly completed form **CANNOT SUE THEIR PHYSICIAN (OR PHYSICIAN'S DELEGATE) FOR FAILURE TO OBTAIN INFORMED CONSENT** for the predictive genetic test being offered. All other causes of action are preserved.

When completed, a copy of the consent form is to be given to the patient
and the original placed in the patient's medical record.

**Model Informed Consent Form for
Prenatal Predictive Genetic Testing For An Adult Onset Condition Not Expected
to Cause Signs or Symptoms Before the Age of Majority**

Before you agree to be tested, make sure you understand all the information on this form. The purpose of the consent process is to help you consider all the pros and cons of testing, so you can make an informed decision.

By signing the "accept testing" line at the end of this form, you agree that you have received a full explanation of the test and that you have given your informed consent to the test.

If you change your mind, tell your health care provider immediately. The test process will be stopped, but there may still be charges for the work done before you notified your provider.

By signing the "decline testing" line at the end of this form, you show that you have decided not to be tested at this time.

When you sign a properly completed form, you waive any claims against your physician or physician's delegate for failure to obtain informed consent concerning this test. No other claims are waived. You will be given a copy of the completed form.

What is the nature and purpose of this test?

The name of the test is: _____

We are trying to learn whether your fetus has the following (which is or is an indicator of an adult onset disorder not expected to cause signs or symptoms before the age of majority):

_____ changes in chromosome structure or number

_____ gene mutation(s) causing: _____
(name of condition)

_____ evidence of a physical defect called: _____

_____ evidence of infection by: _____
(agent)

_____ other: _____

You have been asked to consider this test because of:

_____ chance for any chromosome problem 1 in _____

_____ chance for _____ 1 in _____

_____ risk for inherited condition called _____

_____ other _____

The lab uses a sample of:

_____ amniotic fluid
_____ chorionic villi
_____ fetal blood from the umbilical cord
_____ fetal biopsy
_____ other: _____

The test is conducted on:

_____ Chromosomes
_____ DNA or RNA
_____ Protein or metabolites (includes amniotic fluid measurements such as AFP)
_____ Infectious particles
_____ Other: _____

Your sample will be sent to (reference laboratory or laboratories):

1. _____
2. _____

The laboratory cost is about \$ _____. There will be added charges for the sampling procedure, processing, shipping and handling. Insurance may not cover your costs.

We usually get results in: _____ days / weeks.

How effective is this test? What are its limitations?

The field of genetics changes very fast. The information we are giving you today reflects current understanding, but it may be modified even a few months from now. If your test results become important for a decision in the distant future, you should check with your health care provider to make sure your information is up to date.

In every medical test there is a small chance for error. Sometimes samples are damaged in transport. Sometimes they are labeled wrong. Sometimes equipment doesn't run correctly or the results are not interpreted correctly. The chance this will happen, and escape detection, is small because safeguards are built in. Because genetic tests are so specialized, they may have other limits as well.

For this particular test:

The accuracy rate is _____ for _____ (test component)
The accuracy rate is _____ for _____ (test component)
The accuracy rate is _____ for _____ (test component)

Rare events which may lead to an incomplete or false result include:

- not enough sample, or wrong kind of tissue

- sample may be taken too early or too late (e.g. protein levels, infectious particles)
- sample culture may not grow
- sample may not truly reflect the fetus
 - maternal cells may be mixed with the fetal cells
 - sample may represent a unique cell population (e.g. placental mosaicism)
- there may be atypical chromosomal inheritance (e.g. uniparental disomy)
- there may be harmful chromosome changes too small to be seen
- there may be other genetic errors we cannot detect using this test
- Polymerase Chain Reaction (PCR), if used, may not work correctly or may enhance the wrong DNA / RNA segment

_____ We will be relying on the results of a linkage study previously conducted on your family. The accuracy of the test results will depend on:

- the accuracy of the linkage studies
 - the distance between the markers and the actual gene change
 - the accuracy of the medical diagnosis
 - the accuracy of the family history
- the accuracy of the parentage reported for this pregnancy
(If you have used donor eggs or sperm, or there is a chance that someone else is the father, tell your health care provider BEFORE the test is done. We are committed to your confidentiality as well as to your health care.)

_____ There is a chance that the test will be "uninformative" or inconclusive -- that is, we won't know any more after testing than we knew before testing.

_____ The test results will not be reliable if the original diagnosis is not accurate. Sometimes two or more genes cause similar symptoms. If we perform tests on one gene and a different gene is at fault, the test results will be meaningless.

If your fetus doesn't have the changes we test for:

- _____ a confirmatory test is recommended after birth.
- _____ your child isn't expected to get _____ (name of condition).
- _____ your child could still have _____ (name of condition), even though we can't detect it.
- _____ the laboratory results will include your child's residual (remaining) risks for the condition.
- _____ your child could get a similar condition from a different gene or from other causes.

If your fetus has one of the changes we test for:

- _____ a confirmatory test is recommended after birth.

_____ your child will have an increased risk for specific health problems, which we will discuss with you after the test results are back.

_____ the chance your child will get the condition depends on the size of the gene expansion. Below _____ repeats, there is no chance of getting the condition.

Between _____ is a gray zone. Some people get symptoms and others don't.

Above _____ repeats, there is a high probability (_____%) of getting symptoms.

_____ Other _____

What are the medical risks and benefits of testing?

We will tell you about the specific risks pertaining to your procedure.

Here are some common, manageable consequences of fetal sampling:

- mild to moderate discomfort during the sampling
- tenderness for a few days at the sampling site
- mild cramping (like menstrual cramps) after the procedure
- light vaginal spotting or bleeding
- restrictions on activity following the procedure
- Rho-gam injection is given if the mother is Rh negative

Here are some uncommon, serious consequences of fetal sampling:

- reaction to anesthetic, if used
- infection at the sampling site; reaction to medicines used to treat infection
- severe cramping or induction of labor; reaction to medicines used to stop labor
- bright or heavy bleeding from the vagina
- leakage of amniotic fluid from the vagina
- fetal injury or death due to complications of sampling procedure
- maternal injury or death due to complications of sampling procedure
- chance for a spontaneous miscarriage at this stage of pregnancy: _____
- added miscarriage risk due to the sampling procedure: _____

If the test shows an increased risk for the condition(s):

_____ there are treatments during pregnancy which may improve your child's outcome.

_____ the condition may prompt changes in your delivery plans (time, place, mode of delivery).

_____ there are treatments after birth which may improve your child's outcome.

_____ currently we don't know of anything which will delay or prevent symptoms for your child but you can use this time to become better informed.

_____ this information may be important for the care of other family members.

_____ other _____

If the test does not show an increased risk for the condition(s):

_____ your child isn't expected to get _____ (name of condition).

_____ your child's risk for _____ may still be higher than average.

_____ your child will still have the "background" or "population" risk for _____

_____ other _____

What about emotional risks and benefits?

People's reactions to prenatal testing are highly personal. Here are some of the emotions parents have reported:

loss or restoration of the sense of a normal pregnancy

pressure to make a quick decision about testing

concern about the religious aspects of testing

satisfaction and peace when the decision reflects one's true preference

anxiety while waiting for results

anger and guilt if the mother or fetus is harmed by the testing process

a sense of isolation, especially around other pregnant women

uncertainty about what to do if results are not favorable

reduced or increased anxiety about personal health, the pregnancy, the child's future

a sense of guilt if one has passed a harmful gene or chromosome change

grief at the loss of the hoped-for, unaffected child

altered self-image

peace of mind in knowing what's likely to come

a sense of being able to plan for the future

a sense that one has done all one can to assure a good outcome

satisfaction or dissatisfaction with the decision to be tested

relief when results are reassuring

The majority of women who undergo prenatal testing get reassuring news.

For people whose fetuses have an increased risk of having an adult onset genetic condition, prenatal testing speeds up the diagnostic process and the adjustment process. You just find out sooner. But you may also be faced with some choices you would not otherwise have considered. Some people would rather not know in advance.

People whose results are inconclusive have the most difficult time. After reaching the decision to be tested, they still don't have an answer. They may feel frustrated and helpless. It often takes them longer to regain their balance.

Genetic test results may throw family relationships into doubt. If the patterns of a parent and child don't match, family secrets (adoption, donor eggs or sperm, infidelity) might be brought to light. THERE ARE SEVERAL GENETIC REASONS why a person's gene

patterns might not match that of their parents. THIS IS NOT A PATERNITY TEST. If you have any questions about this, please ask someone on the medical team BEFORE you are tested. We are committed to your privacy as well as to your medical care.

What about social risks and benefits?

Here are some social issues people consider, regarding testing and the condition itself:
concerns about discrimination in employment or insurance for the mother or the child
concerns about privacy of medical information
concerns about informing at-risk family members
concerns about altered roles and family dynamics (people may treat you differently)
concerns about disapproval by family, friends or community of faith
concerns about narrowed or expanded options in career, family or other arenas
the cost of the test, or the cost of managing the condition, versus other expenses

Genetic test results, like all confidential medical information, can be released only with your written consent. However, if a potential employer or insurer demands that you release your medical records and you refuse, you can be turned down for the job or the insurance. If you have health insurance, your contract allows the insurer access to your own medical records and those of your dependents, even for the medical care it doesn't cover.

No one is sure whether discrimination on the basis of prenatal test results is a significant risk. There are laws prohibiting employment discrimination, but discrimination is difficult to prove and the laws are hard to enforce. Health insurance is a major expense for employers, and many companies "self-insure" which means they have access to their employees' and dependents' health information.

There are laws prohibiting discrimination in health insurance because of presymptomatic conditions. If you have health insurance and your child has a birth defect, your health insurance company cannot refuse to cover the child. But it may not cover all the procedures your child will need, and your child may not be able to get other forms of insurance.

If you have a family history of a genetic condition, you may encounter discrimination based upon your family history alone. A favorable test result may reduce your risk of discrimination.

Most people decide for or against testing by asking themselves whether the medical and emotional benefits and costs of knowing outweigh the medical and emotional benefits and costs of not knowing.

What other options do I have?

Nebraska law says no employer or insurer can require you to have a prenatal genetic test.

_____ You can choose not to be tested.
_____ You can be monitored for symptoms rather than seeking a genetic test.
_____ You can take a different type of test, called _____
The major differences between this test and the test we've been talking about are: _____

_____ You can decide not to be tested now, and test your child later if medically indicated.

_____ You can decide not to be tested now, and let your child decide whether to be tested when he or she becomes an adult.

_____ Other: _____

Additional issues

If there are other issues not covered by the discussion above, write them here.

ACCEPT TESTING

After a full discussion of the risks, benefits and alternatives, I agree to be tested.

For a short time after testing, the lab will keep any remaining sample in case the test must be repeated. After that, the lab may destroy the sample, or it may remove all identifying information and use the sample for research. Two additional options are storage of the sample for your future use (for a fee), or participation in research as an identified subject.

_____ I want to store the sample for my future use. I will be charged for this.

_____ I am willing to be contacted if research options are available, and I will decide whether to participate.

I want to receive my results:

_____ in the health care provider's office. Here are the agreed details:

date and time of appointment _____

address _____

_____ the office will call me when the results are back, and make an appointment.

_____ by phone. Here are the agreed details:

phone number _____

time of day _____

day of week or date of call _____

_____ if I don't answer, leave a message and I'll call you back.

_____ in the mail, addressed as follows:

_____ other: _____

_____ If these arrangements must be changed, please call me at: _____

Please be aware that some testing protocols specify the method of giving results and your doctor may be obligated to honor those protocols.

Patient or patient representative

Date

I have discussed the contents of this form with the patient or patient's representative and have answered his or her questions.

Physician or physician's delegate

Date

DECLINE TESTING

After a full discussion of the risks, benefits and alternatives, I choose not to be tested at this time.

Patient or patient representative

Date

I have discussed the contents of this form with the patient or patient's representative and answered his or her questions.

Physician or physician's delegate

Date

Contacts

If you think of questions after the visit, call this person:

DEFINITIONS & USE OF THE *MODEL* INFORMED CONSENT FORM FOR PREDICTIVE GENETIC TESTING

Nebraska law requires written, informed consent from the patient or the patient's representative before a predictive genetic test is ordered. For purposes of this requirement:

Genetic test means analysis of human DNA, RNA, chromosomes, epigenetic status, and those tissues, proteins and metabolites used to detect heritable or somatic disease-related genotypes or karyotypes for clinical purposes. Tests of tissues, proteins and metabolites are included only when generally accepted in the scientific and medical communities as being specifically determinative of a heritable or somatic disease-related genetic condition. *Genetic test does not include a routine analysis, including a chemical analysis, of body fluids or tissues unless conducted specifically to determine a heritable or somatic disease-related genetic condition. Genetic test does not include a physical examination or imaging study.*

Patient representative includes:

- (1) the custodial parent or guardian of an unemancipated minor¹, except that:
 - (a) a minor who is pregnant has authority to give informed consent or refusal for pregnancy related medical procedures unless, due to physical or intellectual limitations, she is unable to make and communicate a considered judgment about her medical care; and
 - (b) the minor parent of a child has authority to give informed consent or refusal for the medical care of his or her child unless the parent, due to physical or intellectual limitations, is unable to make and communicate a considered judgment about the child's medical care.
- (2) the legal guardian, attorney in fact appointed by a Durable Power of Attorney for Health Care, or other adult who has a history of assuming decisional authority for a patient who, due to physical or intellectual limitations, is unable to make and communicate a considered judgment about his or her medical care.

Physician delegate means an individual acting under the delegated authority of a physician to perform a selected act, task or function, and who understands and is qualified to provide the information required in the written informed consent.

Predictive genetic test means a genetic test for an otherwise undetectable genotype or karyotype relating to the risk for developing a genetically related disease or disability, the results of which can be used to substitute a patient's prior risk based on population data or family history with a risk based on genotype or karyotype. *Predictive genetic test does not include diagnostic testing conducted on a person exhibiting clinical signs or symptoms of a possible genetic condition. Predictive genetic testing does not include gamete testing, preimplantation diagnosis, or prenatal genetic diagnosis, unless the prenatal genetic diagnosis is conducted for an adult-onset condition not expected to cause clinical signs or symptoms before the age of majority. For predictive prenatal genetic testing, a different model form is provided.*

This consent form is intended for patients who have no clinical evidence of the condition for which testing is proposed. It is intended only for clinically accepted tests, not for research protocols, for which an IRB-approved consent form should be used. It may be used for all family members in linkage studies, whether or not they are at risk for the condition. This form is not intended for prenatal testing, diagnostic testing in a symptomatic patient, or carrier testing for recessive conditions.

When properly completed and signed, this form gives evidence that the physician (or physician delegate) and the patient (or patient representative) have thoroughly discussed the risks, benefits and limitations of the proposed test. Patients who have signed this properly completed form CANNOT SUE THEIR PHYSICIAN (OR PHYSICIAN'S DELEGATES) FOR FAILURE TO OBTAIN INFORMED CONSENT for the genetic test being offered. All other causes of action are preserved.

When completed, a copy of the consent form is to be given to the patient
and the original placed in the patient's medical record.

¹ Note however, that predictive testing of a minor child is RARELY APPROPRIATE unless the results of the test will alter medical management before the child reaches the age of majority. This limitation is consistent with guidelines promulgated by professional groups, e.g. ASHG/CMG Report "Points to Consider: Legal, Ethical and Psychosocial Implications of Genetic Testing in Children and Adolescents" Am J. Hum. Genet. 57-1233-1241 (1995); AAP Policy Statement "Molecular Genetic Testing in Pediatric Practice; A Subject Review (RE0023) Pediatr 106(6); 1494-1497 (2000). Use of this form will not protect against possible liability for improper testing of a minor for adult onset conditions

Model Informed Consent Form for Predictive Genetic Testing

Before you agree to be tested, make sure you understand all the information on this form.

By signing the "accept testing" line at the end of this form, you agree that you have received a full explanation of the test and that you have given your informed consent to the test.

If you change your mind, tell your health care provider immediately. The test process will be stopped, but there may still be charges for the work done before you notified your provider.

When you sign a properly completed form, you waive any claims against your physician or physician's delegate for failure to obtain informed consent concerning this test. No other claims are waived. You will be given a copy of the completed form.

What is the nature and purpose of this test?

The name of the test is: _____

_____ We are trying to learn whether you are likely to get _____
because of your genetic status. *(name of condition)*

_____ Your sample is needed to help other family members learn whether they are
likely to get _____ because of their genetic
status. *(name of condition)*

The lab uses a sample of:

_____ blood
_____ cells gathered from your mouth
_____ skin
_____ muscle
_____ sweat
_____ other body fluid: _____
_____ other: _____

The test is conducted on:

_____ chromosomes
_____ DNA or RNA
_____ protein or metabolites
_____ other: _____

Your sample will be sent to this laboratory: _____

The laboratory cost is about \$ _____. There may be added charges for the sampling procedure, processing, shipping and handling. Insurance may not cover your costs.

We usually get results in _____ days / weeks.

How effective is this test? What are its limitations?

The field of genetics changes very fast. The information we are giving you today reflects the current understanding, but it may be quite different even a few months from now. If you will be making decisions based on these test results in the future, you should first check with your health care provider to make sure your information is up to date.

In every medical test there is a small chance for error. Sometimes samples are damaged in transport. Sometimes they are labeled wrong. Sometimes equipment doesn't run correctly or the results are not interpreted correctly. The chance this will happen, and escape detection, is small because safeguards are built in. Because genetic tests are so specialized, they may have other limits as well.

For this particular genetic test:

_____ we will rely on linkage analysis.

- The accuracy of the test will depend on:
 - correct genetic diagnosis
 - accurate medical histories of family members
 - complete, accurate information about the family tree, including:
 - adoptions
 - donor eggs or sperm
 - other possible differences in the family bloodlines
(we will respect the confidentiality of sensitive information)
 - distance between the genetic marker and the actual genetic change

- We give people their own test results. We don't share information about relatives unless they give us permission. Even so, you may learn things about your relatives when you learn your own genetic status. It is important to respect their privacy and confidentiality, just as you want them to respect yours.

_____ Your results will not have any medical significance. They will be used only to establish the family's genetic patterns.

_____ there is a chance that the test will be "uninformative" or inconclusive -- that is, we won't know any more after testing than we knew before testing.

_____ the test results will not be reliable if the original diagnosis is not accurate. Sometimes two or more genes cause similar symptoms. If we perform tests on one gene and a different gene is at fault, the test results will be meaningless

If you don't have the genetic changes we test for:

_____ you are not expected to get _____
(name of condition).

_____ you could still get _____ (name of condition), or
be a carrier, because of an error in a different part of the gene or chromosome.
_____ you could still get a similar condition from a different gene or from other causes.

If you have the genetic changes we test for:

_____ you have an increased risk for specific health problems, which we will discuss
with you after the test results are back.

_____ the chance you will get the condition depends on the size of your gene
expansion. below _____ repeats, there is no chance of getting the condition.
between _____ is a gray zone. Some people get symptoms and
others don't.

above _____ repeats, there is a high probability (_____%) of getting
symptoms.

_____ you could pass the genetic change to your children.

_____ other _____

What are the medical risks and benefits?

The medical risks of taking the sample vary with the type of procedure required. When blood is drawn, there may be temporary discomfort, bleeding (especially if you bleed easily or take a blood thinner), and bruising. If you have a biopsy, you may have a reaction to the anesthetic, bleeding, an infection, scarring or poor wound healing at the sampling site, and discomfort for a few days. If there are other risks, we will discuss them with you and we may ask you to sign a separate consent for the sampling procedure itself.

If the test shows an increased risk for the condition:

_____ you won't need any more tests to figure out the diagnosis.

_____ you may need more testing to monitor and manage your condition.

_____ there are things you can do to delay the onset or reduce the severity of
symptoms.

_____ currently we don't know of anything which will delay or prevent symptoms.

_____ this information may be important for the care of other family members.

_____ other _____

If the test does not show an increased risk for the condition:

_____ you are not expected to get _____
(name of condition).

_____ your risk may still be higher than average, and your children may still be at risk.

_____ you will still have the "background" or "population" risk for _____
(name of condition)

_____ your doctor will not need to see you as often for monitoring.

_____ this information may be important for the care of other family members.
_____ other _____

What about emotional risks and benefits?

Here are some of the emotions people report after testing, regardless of their results:

*letdown after the intensity of the testing process
increased or reduced anxiety about personal health
increased or reduced anxiety about relatives, especially children
a sense of guilt, especially if the tested person is spared and siblings are not
depression, sometimes serious, even if the results are favorable
uncertainty about what to do, especially when results are surprising
altered self-image
peace of mind in knowing what's likely to come
a sense of being able to plan for the future
relief on learning results
satisfaction or dissatisfaction with the decision to be tested*

Studies show that emotional reactions can be intense in the first several weeks after testing. Within a few months after testing, people generally report better emotional health than they had before testing. People who choose not to be tested have the same emotional health that they had before.

People whose results are inconclusive have the most difficult time. After reaching the decision to be tested, they still don't have an answer. They may feel frustrated and helpless. It often takes them longer to regain their balance.

Genetic test results may throw family relationships into doubt. If the patterns of a parent and child don't match, family secrets (adoption, donor eggs or sperm, infidelity) might be brought to light. THERE ARE SEVERAL GENETIC REASONS why a person's gene patterns might not match that of their parents. THIS IS NOT A PATERNITY TEST. If you have any questions about this, please ask someone on the medical team BEFORE you are tested. We are committed to your privacy as well as to your medical care.

Professional help is available to come to terms with the results of genetic testing.

If there are other major stresses in your life right now, or you feel like you don't have the support you need, this may not be a good time to be tested. You can postpone testing until later.

What about social risks and benefits?

Here are some social issues people consider before testing:

*increased or decreased risk of discrimination in employment and insurance
concerns about privacy of medical information*

concerns about altered roles and family dynamics (people may treat you differently)
concerns about telling other family members who may be at risk
concerns about narrowed or expanded options in career, family or other arenas
the cost of the test versus other expenses

Genetic test results, like all confidential medical information, can be released only with your written consent. However, if a potential employer or insurer demands that you release your medical records and you refuse, you can be turned down for the job or the insurance. If you have health insurance, your contract allows the insurer to have access to all of your medical records, even the services it doesn't pay for.

No one is sure whether discrimination on the basis of predictive test results is a significant risk. There are laws prohibiting employment discrimination, but discrimination is difficult to prove and the laws are hard to enforce. Health insurance is a major expense for employers, and many companies "self-insure" which means they have access to their employees' health information.

There are laws prohibiting discrimination in health insurance because of presymptomatic conditions, but the laws don't cover other types of insurance. Withholding relevant medical information in an insurance application is fraudulent. Some people think presymptomatic testing isn't worth the risk of possible discrimination in employment or insurance.

If you have a family history of a genetic condition, you may encounter discrimination based upon your family history alone. A favorable test result may reduce your risk of discrimination.

Most people decide for or against testing by asking themselves whether the medical and emotional benefits of knowing outweigh the medical and emotional costs of not knowing.

What other options do I have?

Nebraska law says no employer or insurer can require you to have a predictive genetic test.

- ☐ You can choose not to be tested.
- ☐ You can wait until you feel better prepared for the test.
- ☐ You can wait for more accurate or thorough tests to be developed.
- ☐ You can be monitored for symptoms rather than seeking a genetic test.
- ☐ You can take steps to reduce the risk of the condition, whether or not you carry the gene.
- ☐ You can have surgery.
- ☐ You can modify your lifestyle.
- ☐ You can take medicine.

_____ Other _____
_____ You can take a different type of test, called _____
The major differences between this test and the test we've been talking about are: _____

Additional issues

If there are other issues not covered by the discussion above, write them here.

ACCEPT TESTING

After a full discussion of the risks, benefits and alternatives, I agree to be tested.

For a short time after testing, the lab will keep any remaining sample in case the test must be repeated. After that, the lab may destroy the sample, or it may remove all identifying information and use the sample for research. Two additional options are storage of the sample for your future use (for a fee), or participation in research as an identified subject.

_____ I want my sample to be stored for my future use. I will be charged for this.

_____ I am willing to be contacted if research options are available, and I will decide whether to participate.

I want to receive my results:

_____ in the health care provider's office. Here are the agreed details:

date and time of appointment: _____

address: _____

_____ the office will call me when the results are back, and make an appointment.

_____ by phone. Here are the agreed details:

phone number: _____

time of day: _____

day of week or date of call: _____

_____ in the mail, addressed as follows:

_____ other: _____

_____ If these arrangements must be changed, please call me at: _____

Please be aware that some testing protocols specify the method of giving results and your doctor may be obligated to honor those protocols.

Patient or patient representative

Date

I have reviewed the contents of this form with the patient or patient's representative and answered his or her questions.

Physician or physician delegate

Date

DECLINE TESTING

After a full discussion of the risks, benefits and alternatives, I choose not to be tested at this time.

Patient or patient representative

Date

I have reviewed the contents of this form with the patient or patient's representative and answered his or her questions.

Physician or physician's delegate

Date

DEFER DECISION

After a full discussion of the risks, benefits and alternatives, I choose to consider my options for a time before deciding whether to be tested.

Patient or patient's representative

Date

I have reviewed the contents of this form with the patient or patient's representative and answered his or her questions.

Physician or physician's delegate

Date

Contacts

If you think of questions after the visit, call this person:

